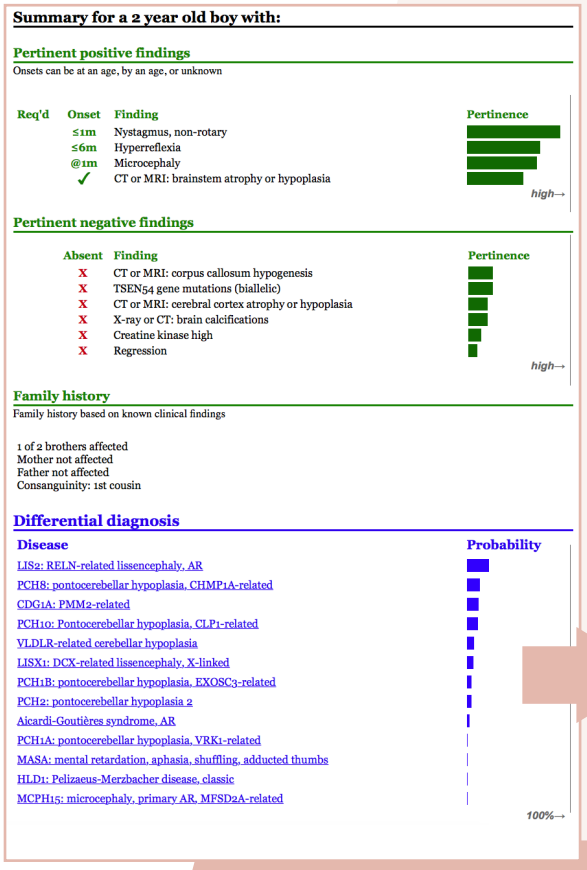
Loss of Heterozygosity

Find the pertinent genes and likely diagnosis in seconds



Summary for a 2 year old boy with:

Pertinent positive findings

Onsets can be at an age, by an age, or unknown

Req'd	Onset	Finding	Pertinence
	≤1m	Nystagmus, non-rotary	
	≤6m	Hyperreflexia	
	@1m	Microcephaly	
✓		CT or MRI: brainstem atrophy or hypoplasia	

high→

Pertinent negative findings

Absent	Finding	Pertinence
X	CT or MRI: corpus callosum hypogenesis	
X	TSEN54 gene mutations (biallelic)	
X	CT or MRI: cerebral cortex atrophy or hypoplasia	
X	X-ray or CT: brain calcifications	
X	Creatine kinase high	
X	Regression	

high→

Family history

Family history based on known clinical findings

1 of 2 brothers affected
Mother not affected
Father not affected
Consanguinity: 1st cousin

Differential diagnosis

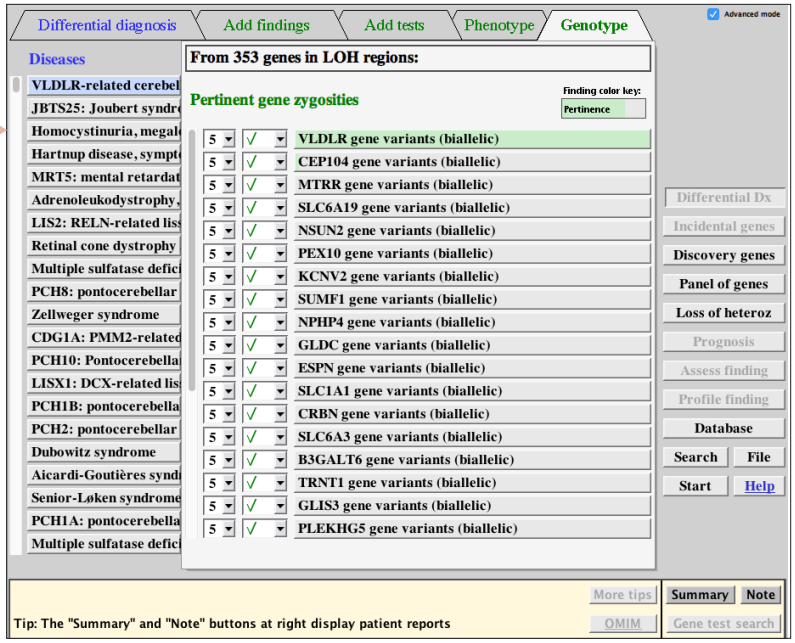
Disease	Probability
LIS2: RELN-related lissencephaly, AR	
PCH8: pontocerebellar hypoplasia, CHMP1A-related	
CDG1A: PMM2-related	
PCH10: Pontocerebellar hypoplasia, CLP1-related	
VLDLR-related cerebellar hypoplasia	
LISX1: DCX-related lissencephaly, X-linked	
PCH1B: pontocerebellar hypoplasia, EXOSC3-related	
PCH2: pontocerebellar hypoplasia 2	
Aicardi-Goutières syndrome, AR	
PCH1A: pontocerebellar hypoplasia, VRK1-related	
MASA: mental retardation, aphasia, shuffling, adducted thumbs	
HLD1: Pelizaeus-Merzbacher disease, classic	
MCPH15: microcephaly, primary AR, MFSD2A-related	

100%→

Key Features

- Automatically correlates the list of patient findings with the genes in chromosomal regions
- Identifies and ranks the **genes by pertinence** for this patient
- Enables a quick call to the ordering physician to focus on additional findings that help reach a conclusion, relying on the **"useful findings"** and **"useful tests"** features

CONSULT on genes in regions of loss of heterozygosity



Key Benefits

Focused on your patient. Uses your patient's pertinent positive and negative findings to generate a list of candidate genes in order of pertinence

Fast. Analysis in seconds, enables useful discussion with referring physician, when needed

Accurate. Reduces errors and inconsistencies

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